

## Molecular Pathology Tests

### Cytogenetics

#### Chromosome Studies

##### Postnatal:

- Peripheral blood
- Fibroblasts

##### Products of Conception:

- CVS (chorionic villi)
- Fetal tissue
- Placenta
- Skin
- Bone

##### Cancer:

- Bone Marrow
- Leukemic blood
- Lymph node
- Fatty tumors
- Renal tissue
- Solid tumors

##### Constitutional Chromosomal Microarray

- Whole Genome microarray (SNP array)

##### Constitutional FISH:

- X cen on PB
- Y cen on PB
- Yqh on PB
- SRY on PB
- X/ST on PB

### Molecular Genetics and Genomics

- ACTA2 exon sequencing
- Alpha-1-antitrypsin genotyping (S and Z alleles)
- Cystic Fibrosis NGS
- DNA fingerprinting for identity (STR;PCR)
- Factor V Leiden (PCR)
- Fragile X syndrome (*FMR1*) DNA analysis (PCR)
- HFE* [Hereditary Hemochromatosis] (PCR)
- MTHFR* (PCR) (C677T,AIZ98C)
- NOD2 exon sequencing
- Progenitor cell engraftment monitoring (STR;PCR)
- Prothrombin G20210A (PCR)
- SERPINA1 sequencing
- TGFBR2 exon sequencing

### Molecular Hematopathology

- Alpha thalassemia (*HBA1/2*) gene deletions

- B-cell clonality (*IGH* + *IGK* BIOMED-2 PCR)
- BCR-ABL Qualitative Multiplex RT-PCR
- CEBPA* mutation analysis
- Diffuse Large B-Cell Lymphoma
- T-cell clonality (TCRB + TCRG BIOMED-2 PCR)
- IGH* PCR (BIOMED-2 Primers)
- IGK* PCR (BIOMED-2 Primers)
- KIT* AML mutation analysis
- KIT* D816V (PCR)
- MPL* mutation analysis
- MYD88* L265P mutation analysis
- TCRB* PCR (BIOMED-2 Primers)
- TCRG* PCR (BIOMED-2 Primers)
- p210 *BCR/ABL1*( RT-PCR, quantitative)
- p190 *BCR/ABL1*( RT-PCR, quantitative)
- BCR/ABL* Kinase Domain Mutation Analysis
- PML/RARA* RT-PCR, qualitative
- Nucleophosmin gene (*NPM1*) mutation analysis
- FLT3* Gene Mutations\*\*
- JAK2* V617F mutation (PCR)
- JAK2* exons 12-15 sequencing

#### Fluorescence in-situ hybridization

- 5q
- ALK*
- BCL2*
- BCL6*
- BCR/ABL1*
- BIRC3(API2)/MALT1*
- CBFB/MYH11* (inv 16)
- CLL (13q,11q, 17p,+12)
- ETV6/RUNX1 (TEL/AML1)*
- FGFR1*
- IGH*
- IGH/BCL2*
- IGH/CCND1*
- IGH/MALT1*
- IGH/MYC*
- MALT1(18q21)*
- MLL (KMT2A)*
- MYC*
- Myelodysplasia (-5/5q, -7/7q,+8,-20q)
- Plasma cell myeloma [13q, IGH, TP53, t(11,14), t(4;14), t(14;16)]
- PDGFRA*

\* Validated, soon to be implemented

\*\* Performed at LabPMM

Continued on back

*PDGFRB*  
*PML/RARA*  
*RARA*  
*RUNX1/RUNX1T1 (AML1/ETO)*  
 Trisomy 4, 10 and 17

## Molecular Microbiology

Affirm (for vaginitis)  
 BK virus (FISH)  
 BK virus PCR, quantitative  
*Bordetella pertussis* (NAAT)  
*Chlamydia trachomatis* (NAAT)  
*Clostridium difficile* PCR  
 CMV detection (quantitative CMV PCR)  
 Dimorphic fungi (probe analysis)  
 EBV detection (quantitative EBV PCR)  
 Enterovirus detection (RT-PCR)  
 HBV detection (quantitative HBV PCR)  
 HCV genotyping  
 HCV viral load (quantitative HCV RT-PCR)  
 HIV DNA PCR  
 HIV viral load (quantitative HIV RT-PCR)  
 HPV DNA (high-risk types)  
 HSV PCR (for CSF)  
 Influenza A/Influenza B/RSV (multiplexed RT-PCR)  
*Legionella pneumophila* (PCR)  
*Mycobacterium sp.* identification (pyrosequencing and probe analysis)  
*Mycobacterium tuberculosis* vs. NTM PCR  
*Neisseria gonorrhoeae* (NAAT)  
*Nocardia* and other aerobic actinomycetes identification (pyrosequencing)  
*Pneumocystis jirovecii* (PCR)  
 Respiratory Virus Multiplex PCR  
*S. aureus* (MRSA/MSSA) PCR (screens from nasal swabs)  
*Streptococcus* Group A, PCR  
*Streptococcus* Group B PCR

## Molecular Oncology

*BRAF* for Hairy Cell Leukemia  
*BRAF* mutational analysis  
*EGFR* mutational analysis  
*FISH* for MYC Angiosarcoma  
*HER2* (ERBB2) with D17S122  
*KRAS* mutational analysis  
*MGMT* promoter methylation

Microsatellite Instability (MSI)  
*PCA3*  
*RET*  
*ROS1*  
 Transthyretin (TTR) exon sequencing  
 VWF exon 28 sequencing

## Fluorescence in-situ hybridization

*1p*  
*19q*  
*ALK* NSCLC FFPET  
*ALK* NSCLC ThinPrep  
*DDIT3* (CHOP)  
*EGFR* (gliomas)  
*EWSR1*  
 FISH for bladder CA recurrence  
*FOXO1A* (FKHR)  
*FUS*  
*HER2* (ERBB2) with *CEP17*  
*MDM2*  
*SS18* (SYT)

## Tests in Development

*ACTA2* exon sequencing\*  
 Bacterial Sequence-Based Identification  
 Beta Catenin (Soft Tissue Tumors)\*  
 Bordetella pertussis by NAAT  
 Cancer Hotspot NGS panel-Colon Cancer\*  
 Calreticulin (*CALR*)\*  
*COL3A1* exon sequencing  
 Constitutional Gene Panel, v1  
 Cystic Fibrosis NGS  
 FISH for Biliary Tract Neoplasia  
 HeartGene Panel\*  
 Mycobacterial Identification/Resistant Determinants (NGS)  
 RT PCR for *EWSR1* (gene rearrangement)  
 RT PCR for *SYT* (gene rearrangement)  
 Solid Tumor SNP chromosomal array  
 Spectral Karyotyping  
 Bone Marrow  
 Blood  
 Cell lines  
*SMAD3* exon sequencing  
*TGFBR1* exon sequencing\*  
*TGFBR2* exon sequencing\*  
 Vascular Panel (*ACTA2*, *COL3A1*, *SMAD3*, *TGFBR1*, *TGFBR2*)\*

\* Validated, soon to be implemented

\*\* Performed at LabPMM